

ABSTRACT

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The present invention relates to a novel human ABCA12 gene as well as cDNAs encoding the novel full and short length ABCA12 proteins. The invention also relates to vectors and recombinants host cells comprising such nucleic acids, nucleotide probes and primers, and means for the detection of polymorphisms and mutations in the ABCA12
10 gene or in the corresponding protein produced by the allelic form of the ABCA12 gene.